The John Walton
MUSCULAR DYSTROPHY
RESEARCH CENTRE
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The Myotubular and
Centronuclear Myopathy
Patient Registry

Myotubular & Centronuclear Myopathy (MTM-CNM) Patient Registry

The MTM -CNM registry is an international patient-initiated registry with over 520 patients registered across 28 countries and available in ten languages. Patient registries are a long-established and valuable way of collecting real-world data, assisting with patient engagement and supporting research into this rare condition.

This registry is responsive to evolving research knowledge into MTM-CNM and has recently been involved in two exciting studies which we'd like to share with you.

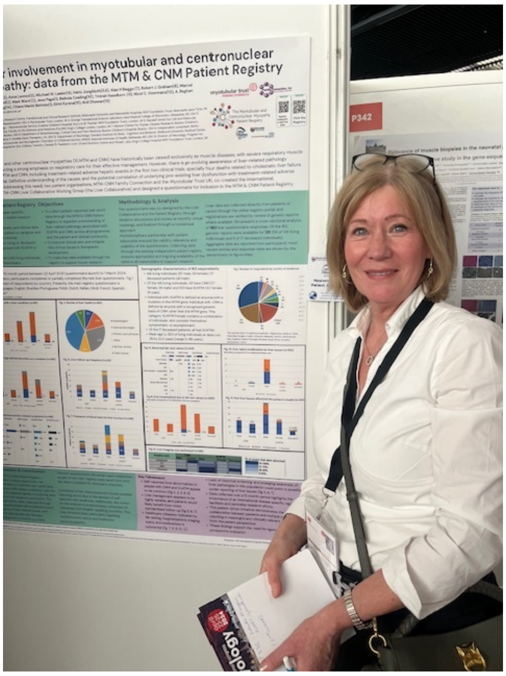
Brain MRI Study

SickKids hospital, Toronto (led by PI - Dr Jim Dowling) is undertaking a research study with the aim of the study being to better understand any changes in the brain for MTM patients. The registry has been asked to partner in this work by helping to collect patient data and the criteria for study selection is as follows:

- male patients (alive & deceased)
- confirmed genetic diagnosis of X-linked MTM (XLMTM)
- previously had brain MRI (imaging) undertaken

Patients registered who meet the above criteria are invited to participate in the study. It's easy to be involved and involves answering a couple of registry questions and the registry then will follow up with the doctor to do the rest. We hope as many of you can contribute to this interesting new research as possible and please do get in touch should you have any questions.

Liver Health Study



Following concerns that liver issues in patients with MTM-CNM were possibly being under-reported, a liver health questionnaire was developed by a group of experts and implemented into the registry in April 2023. We are delighted that over 165 participants have now completed it and the results of this have now been presented at the World Muscle Society, October 2023 and International Congress of Myology, April 2024. Data entered by patients provides a valuable insight into potential liver-related issues in CNM and a better understanding of the number of people impacted. New research is looking into a potential link between diet and liver health and so the registry questionnaire will shortly be updated to include questions on nutrition and diet.

We invite and encourage patients and families to join this international registry and support the collection of real-world evidence to improve our understanding of this condition. Registrations are welcomed from MTM-CNM patients (living and deceased) along with female carriers who are either symptomatic or asymptomatic.

Registration is an easy process via this link and if you have any questions please do not hesitate to contact the registry for support by emailing us at mtmcnmregisty@newcastle.ac.uk

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